



## MYO7A gene

myosin VIIA

### Normal Function

The *MYO7A* gene provides instructions for making a protein called myosin VIIA, which is part of a group of proteins called unconventional myosins. These proteins, which have similar structures, help transport molecules within cells. Myosins interact with actin, a protein that is important for cell movement and shape. Researchers believe that myosins use long filaments of actin as tracks along which to transport other molecules.

Myosin VIIA is made in the inner ear and in the retina, which is the light-sensitive tissue at the back of the eye. In the inner ear, myosin VIIA plays a role in the development and maintenance of hairlike projections called stereocilia. Stereocilia, which are rich in actin, line the inner ear and bend in response to sound waves. This bending motion is critical for converting sound waves to nerve impulses, which are then transmitted to the brain. Stereocilia are also elements of the vestibular system, the part of the inner ear that helps maintain the body's balance and orientation in space. Bending of these stereocilia is needed to transmit signals from the vestibular system to the brain.

In the retina, myosin VIIA is found primarily in a thin layer of cells called the retinal pigment epithelium (RPE). Myosin VIIA probably plays a role in the development and maintenance of this tissue, which supports and nourishes the retina. Research suggests that one function of myosin VIIA is to carry small sacs of pigment (called melanosomes) within the RPE. This pigment is necessary for normal vision. Myosin VIIA is also found in other parts of the retina, where it likely carries additional proteins and molecules that are important for vision.

### Health Conditions Related to Genetic Changes

#### nonsyndromic hearing loss

Researchers have identified several *MYO7A* gene mutations that can cause nonsyndromic hearing loss, which is loss of hearing that is not associated with other signs and symptoms. Mutations in this gene are thought to cause two forms of nonsyndromic hearing loss: DFNA11 and DFNB2.

DFNA11 is inherited in an autosomal dominant pattern, which means only one mutated copy of the *MYO7A* gene in each cell is sufficient to cause the condition. This form of hearing loss begins in childhood, after a child learns to speak (postlingual), and becomes more severe over time.

Most of the mutations that cause DFNA11 alter a single protein building block (amino acid) in myosin VIIA, resulting in an abnormal protein that does not work properly. Other genetic changes delete a small amount of DNA from critical regions of the *MYO7A* gene, which probably changes the structure of the protein. Researchers suspect that the altered protein causes hearing loss by disrupting the growth and organization of stereocilia in the inner ear.

DFNB2 is inherited in an autosomal recessive pattern, which means both copies of the *MYO7A* gene are mutated in each cell. The hearing loss can be postlingual or begin before a child learns to speak (prelingual). Some researchers have suggested that individuals with DFNB2 may actually have Usher syndrome (described below), because some individuals who were thought to have nonsyndromic hearing loss developed retinitis pigmentosa (a vision disorder characteristic of Usher syndrome) later in life. However, other individuals diagnosed with DFNB2 never develop retinitis pigmentosa, and recent studies indicate that DFNB2 and Usher syndrome probably result from different mutations in the *MYO7A* gene.

The mutations that cause DFNB2 alter the structure and function of myosin VIIA, but they probably do not eliminate the protein's function completely. Recent studies found that the protein likely retains enough function in the retina to allow for normal vision, but not enough function in the inner ear to permit normal hearing.

### Usher syndrome

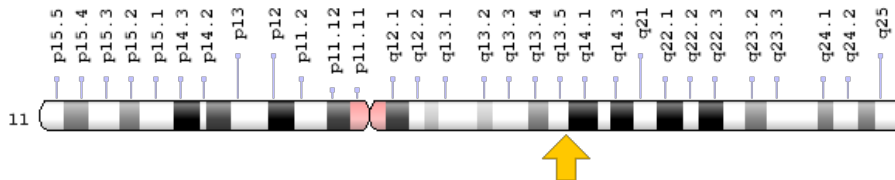
More than 200 mutations in the *MYO7A* gene have been identified in people with Usher syndrome type I, which is characterized by a combination of hearing loss, vision loss, and problems with balance and coordination. Specifically, *MYO7A* gene mutations cause a form of the disorder known as Usher syndrome type IB (USH1B), which accounts for more than half of all cases of Usher syndrome type I.

Many of these genetic changes alter a single protein building block (amino acid) in critical regions of the myosin VIIA protein. Other mutations introduce a premature stop signal in the instructions for making myosin VIIA. Still other mutations insert or delete small amounts of DNA in the *MYO7A* gene. All of these changes lead to the production of a nonfunctional version of myosin VIIA or prevent the production of any of this protein. A lack of myosin VIIA in the inner ear disrupts the normal development and function of stereocilia, which leads to hearing loss and difficulty with balance and coordination. A lack of myosin VIIA in the retina causes retinitis pigmentosa, a condition in which light-sensing cells of the retina gradually deteriorate, resulting in progressive vision loss.

## Chromosomal Location

Cytogenetic Location: 11q13.5, which is the long (q) arm of chromosome 11 at position 13.5

Molecular Location: base pairs 77,128,192 to 77,215,241 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- DFNA11
- DFNB2
- MYO7A\_HUMAN
- myosin VIIA (Usher syndrome 1B (autosomal recessive, severe))
- NSRD2
- USH1B

## Additional Information & Resources

## Educational Resources

- Neuroscience (second edition, 2001): Hair Cells and the Mechanoelectrical Transduction of Sound Waves  
<https://www.ncbi.nlm.nih.gov/books/NBK10867/>
- Neuroscience (second edition, 2001): The Retina  
<https://www.ncbi.nlm.nih.gov/books/NBK10885/>
- The Cell: A Molecular Approach (second edition, 2000): Unconventional Myosins  
<https://www.ncbi.nlm.nih.gov/books/NBK9961/#A1804>

### GeneReviews

- Deafness and Hereditary Hearing Loss Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1434>
- Usher Syndrome Type I  
<https://www.ncbi.nlm.nih.gov/books/NBK1265>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MYO7A%5BTIAB%5D%29+OR+%28myosin+VIIA%5BTIAB%5D%29%29+OR+%28%28USH1B%5BTIAB%5D%29+OR+%28DFNA11%5BTIAB%5D%29+OR+%28DFNB2%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### OMIM

- MYOSIN VIIA  
<http://omim.org/entry/276903>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_MYO7A.html](http://atlasgeneticsoncology.org/Genes/GC_MYO7A.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=MYO7A%5Bgene%5D>
- HGNC Gene Family: A-kinase anchoring proteins  
<http://www.genenames.org/cgi-bin/genefamilies/set/396>
- HGNC Gene Family: Deafness associated genes  
<http://www.genenames.org/cgi-bin/genefamilies/set/1152>
- HGNC Gene Family: FERM domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/1293>
- HGNC Gene Family: Myosins, class VII  
<http://www.genenames.org/cgi-bin/genefamilies/set/1102>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=7606](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7606)
- Leiden Open Variation Database: MYO7A Gene Mutations  
[https://research.cchmc.org/LOVD2/home.php?select\\_db=MYO7A](https://research.cchmc.org/LOVD2/home.php?select_db=MYO7A)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/4647>

- RetNet: Summaries of Genes and Loci Causing Retinal Diseases: MYO7A  
<https://sph.uth.edu/retnet/disease.htm#11.206d>
- The Hereditary Hearing Loss Homepage  
<http://hereditaryhearingloss.org/>
- UniProt  
<http://www.uniprot.org/uniprot/Q13402>

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